

1 : NM\_000787 . Homo sapiens  
dopam...[gi:4503260]

PubMed, Protein, Related Sequences

LinkOut

LOCUS NM\_000787 2725 bp mRNA PRI 19-MAR-1999  
 DEFINITION Homo sapiens dopamine beta-hydroxylase (dopamine beta-monoxygenase) (DBH) mRNA.  
 ACCESSION NM\_000787  
 VERSION NM\_000787.1 GI:4503260  
 KEYWORDS .  
 SOURCE human.  
 ORGANISM Homo sapiens  
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.  
 REFERENCE 1 (bases 1 to 2725)  
 AUTHORS Kobayashi,K., Kurosawa,Y., Fujita,K. and Nagatsu,T.  
 TITLE Human dopamine beta-hydroxylase gene: two mRNA types having different 3'-terminal regions are produced through alternative polyadenylation  
 JOURNAL Nucleic Acids Res. 17 (3), 1089-1102 (1989)  
 MEDLINE 89160241  
 REFERENCE 2 (bases 1 to 2725)  
 AUTHORS Nagatsu,T.  
 TITLE Direct Submission  
 JOURNAL Submitted (14-OCT-1988) Nagatsu T., Department of Biochemistry, Nagoya University, School of Medicine, Nagoya 466, Japan  
 COMMENT REFSEQ: This reference sequence was derived from X13255.  
 see also X13256 for type b mRNA  
 Map data from Craig et al. Cytogenet. Cell Genet. 48:48-50 (1988).  
 PROVISIONAL RefSeq: This is a provisional reference sequence  
 record that has not yet been subject to human review. The final curated reference sequence record may be somewhat different from this one.  
 FEATURES Location/Qualifiers  
 source 1..2725  
 /organism="Homo sapiens"  
 /db\_xref="taxon:9606"  
 /map="9q34"  
 gene 1..2725  
 /gene="DBH"  
 /db\_xref="LocusID:1621"  
 /db\_xref="MIM:223360"  
 CDS 33..1844  
 /gene="DBH"  
 /EC\_number="1.14.17.1"  
 /codon\_start=1  
 /db\_xref="LocusID:1621"  
 /db\_xref="MIM:223360"  
 /product="dopamine beta-hydroxylase (dopamine beta-monoxygenase)"  
 /protein\_id="NP\_000778.1"  
 /db\_xref="GI:4503261"  
 /translation="MREAAFMYSTAVAIFLVILVAALQGSAPRESPLPYHIPLDPEGS  
 LEIWSWNVSYTQEAIHFQLLVRRRLKAGVLFGMSDRGELENADLVLWTDGDTAYFADAW  
 SDQKGQIHLDPOQDYLQVQRTPEGTLFLFKRPFGTCDPKDYLIEDGTVHLVYGLIE"

Figure 1A

EPFRSLEAINGSGLQMGLQRVQLLKPNIPEPELPSDACTMEVQAPNIQIPSQETTYWC  
 YIKELPKGFSRHIIKYEPIVTKGNEALVHMEVFQCAPEMDSVPHFSGPCDSKMKPD  
 RLNYCRHVLAAWALGAKAFYYPEEAGLAFGGPGSSRYLRLEVHYHNPLVIEGRNDSSG  
 IRLYYTAKLRRFNAGIMELGLVYTPVMAIPPRETAFILTGYCTDKCTQLALPPSGIHI  
 FASQLHTHLTGRKVVTVLVRDGREWEIVNQDNHYSPHFQEIRMLKKVSVHPGDVLIT  
 SCTYNTEDRELATVGGFGILEEMCVNYVHYPQTQLELCKTAVDAGFLQKYFHLINRF  
 NNEDVCTCPQASVSQQFTSVPWNSFNRDVLKALYSFAPISMHCNKSSAVRFQGEWNLQ  
 PLPKVISTLEEPTPQCPTSQGRSPAGPTVVSIGGGKG"

siq peptide 33..107  
mat peptide 108..1841  
 /product="dopamine beta-hydroxylase (dopamine  
 beta-monooxygenase)"

BASE COUNT 533 a 901 c 774 g 517 t  
 ORIGIN

1 tcagtcgctg ggccagcctg cccggccca gcatgcggga ggcagccttc atgtacagca  
 61 cagcagtggc catcttcctg gtcatcctgg tggccgact gcagggctcg gctcccccgt  
 121 agagccccct cccctatcac atccccctgg accccggaggg gtccttggag ctctcatgg  
 181 atgtcagcta cacccaggag gccatccatt tccagctctt ggtcggagg ctcaaggctg  
 241 gcgtcctgtt tggatgtcc gaccgtggcg agctttagaa cgcagatctc gtgggtct  
 301 ggaccgatgg ggacactgccc tattttgcgg acgcctggag tgaccagaag gggcagatcc  
 361 acctggatcc ccagcaggac taccagctgc tgcaggtca gaggacccca gaaggccctga  
 421 ccctgctttt caagaggccc tttggcacct gcgacccca ggattacctc attgaagacg  
 481 gcactgtcca cttggtctac gggatcctgg aggagccgtt ccgtcaactg gaggccatca  
 541 acggctcggg cctgcagatg gggctgcaga gggtgcagct cctgaagccc aatatcccc  
 601 aaccggagtt gcccctcagac gctgtcacca tggaggtcca agctccaaat atccagatcc  
 661 ccagccagga gaccacgtac tgggtctaca ttaaggagct tccaaaggcc ttctctccgc  
 721 accacattat caagtacgag cccatcgtca ccaaggccaa tgaggccctt gtccaccaca  
 781 tggaaagtctt ccagtgcgcc cccgagatgg acagcgtccc ccacttcagc gggccctgc  
 841 actccaagat gaaacccgac cgcctcaact actgcccaca cgtgctggcc gcctggggcc  
 901 tgggtccaa ggcattttac tacccagagg aagccggct tgccttggg ggtccaggg  
 961 cctccagata tctccgcctg gaagttcaact accacaaccc actggtgata gaaggacaa  
 1021 acgactcctc aggcatccgc ttgtactaca cagccaagct gcgccgttc aacgcgggaa  
 1081 tcatggagct gggactggtg tacacgccc gtagtggccat tccaccacgg gagaccgcct  
 1141 tcatcctcac tggctactgc acggacaagt gcacccagct ggcactgcct ccctccgg  
 1201 tccacatctt cgcctctcag ctccacacac acctgactgg gagaaaagggt gtcacagtgc  
 1261 tggtccggga cggccggag tggagatcg tgaaccagga caatcaactc agccctcaact  
 1321 tccaggagat ccgcattttg aagaaggctg tgcgggtcca tccgggagat gtgctcatca  
 1381 ctcctgcac gtacaacacg gaagaccggg agctggccac agtggggggc ttcggtatcc  
 1441 tggaggagat gtgtgtcaac tacgtgcact actacccca gacgcagctg gagctctgc  
 1501 agacggctgt ggacgcggc ttctgcaga agtacttca cctcatcaac aggtcaaca  
 1561 acgaggatgt ctgcacctgc cctcaggcgt ccgtgtctca gcagttcacc tctgttccct  
 1621 ggaactcctt caaccgcac gtactgaagg ccctgtacag cttcgcggcc atctccatgc  
 1681 actgcaacaa gtcctcagcc gtccgcctcc agggtaatg gaacctgcag cccctgcccc  
 1741 aggtcatctc cacactggaa gagcccaccc caccagccag ggccgaagcc

Figure 1B

1801 ctgctggccc caccgttgtc agcattggtg gggcaaagg ctgagggggg acctactcct  
1861 ccccttcctc catgctgtcc ctgtggctc acaccggcac tgtgcactct actctgcgac  
1921 gatccccatg gaacagccct gcaacgcccag gatgaagggg ccagaccacg cccctgcctg  
1981 agaccacggt ccaatccagc cttcttcccc cagggtcccc tgcacatggctg agagggtgtg  
2041 ggtgccctgt tgacctaccc tggaccgagt ggaccacgac ctgcgtccatt taaaaccggc  
2101 tgactcagtg cagggacagc ccgcacagtg gtccagggtc cagccctccg ccagccctgt  
2161 tccgcctcac tgggtgtggc ctggcttctg ggacaggcac catgctgggc cggggtgtgg  
2221 aatcaccggg aacgcccccg ccccccggcc gctgctcccg gtgtgcagcg ggtgcgggtg  
2281 ccgcctaaac atttccctgc tgagtggctc gtgtttcaca gtggggcggtc tccctgcgac  
2341 ggaggcagga ccaggcattt agcttagttag agactcgct gggaaattgc tccattcctg  
2401 agtaaacaga tattttcgcc cacctaaagg gaagccctga caacaactat caccaaaaga  
2461 cgaggcggca aagatccagc ggggcttctg ggcgcgggtt ccacgtgggg tggaattatt  
2521 agcaccagct tgcttcttg ccggtggggc cagcgctgaa cagaccgggg tggagtcagg  
2581 gctgtgctt ccgcgtggtt ctgccactta gggagtgtgc cttggggggg ccatttcaca  
2641 ttccctgaccc tcactttct catctgtaaa accaggctga tgccgtgcgg gctaattgagc  
2701 caataaagct cacacttggg ctggc

Figure 1C

## SUMMARY OF DBH ASSOCIATION

Original study	Transmitted	Untransmitted	Chi-sq	p-val
DBHu2	20	21	0.53	0.4658
DBHu1	18	12	1.20	0.2733
DBHp444a	56	41	2.32	0.1278
<b>Total<sup>4</sup></b>				
Replication/DePaulo				
DBHu2	14	8	1.64	0.2008
DBHu1	11	13	0.17	0.6831
DBHp444a	49	38	1.39	0.2383
<b>Total<sup>4</sup></b>				
DBHu2	40	29	1.75	0.1854
DBHu1	29	25	0.30	0.5882
DBHp444a	105	79	3.67	0.0653

## DBH HAPLOTYPE ANALYSIS

Original Study	Transmitted	Untransmitted	Chi-sq	p-val
Allele1 from DBHu2 and u1	37	22	3.81	0.0508
Allele1 from DBHu2 and DBHp444a	45	29	3.40	0.0629
Allele1 from DBHu1 and DBHp444a	43	23	6.06	0.0138
Allele1 from all three SNPs	46	25	6.21	0.0127
<b>Total<sup>4</sup></b>				
Replication/DePaulo				
Allele1 from DBHu2 and u1	18	11	1.69	0.1936
Allele1 from DBHu2 and DBHp444a	32	14	7.04	0.0080
Allele1 from DBHu1 and DBHp444a	31	16	4.79	0.0287
Allele1 from all three SNPs	31	11	9.52	0.0020
<b>Total<sup>4</sup></b>				
Allele1 from DBHu2 and u1	65	33	5.50	0.0190
Allele1 from DBHu2 and DBHp444a	77	43	9.63	0.0019
Allele1 from DBHu1 and DBHp444a	74	39	10.84	0.0010
Allele1 from all three SNPs	77	36	14.88	0.0001